

How Genomics can revolutionise healthcare?

01 January 2023 | Features | By Sanjiv Das

A genomic revolution is taking place in India, where the value of genomic tests is being recognised in the prevention and diagnosis of diabetes, cardiovascular disease, cancer, carrier status, among others. With the power to identify accurate preventive solutions to chart out treatment options for patients, predictive genomic testing is seen as the next major weapon in the arsenal of cutting-edge healthcare tech that can improve patient outcomes without relying on a curative approach. Let's learn more from industry leaders and experts in the field, about leveraging genomics.

A recent study published by Mayo Clinic Proceedings indicates that nearly one in eight people who underwent predictive genomic testing found that they had a genetic risk for a health condition and may be able to manage it better with preventive care. Rising adoption of healthier lifestyles and increasing awareness of new healthcare programmes and advancements are expected to drive increasing demand for predictive genetic testing.

As per a Precedence Research analysis, the global gene therapy market was valued at \$2.99 billion in 2021 and is expected to reach over \$15.68 billion by 2030, which is poised to grow at a registered CAGR of 20.2 per cent from 2022 to 2030.

With a decrease in infectious diseases, there has been a rise in chronic illnesses. Treatments for chronic illnesses are focusing more on an individual's genetic makeup. Though India has 20 per cent of the world's global population, it only contributes to 0.2 per cent of the global genetic database.

Revolutionising diagnosis, prognosis and treatment

Genome sequencing became more prominent and came to the aid during the COVID-19 pandemic where with the help of this technique researchers were able to isolate the SARS-COV-2 virus. This also led to the production of an mRNA vaccine by Moderna within 60 days of the sequence being made available.

There is a growing interest among researchers in this space. Genome sequencing will lead to personalised medicine both from the diagnosis, prognosis and treatment perspective. It could also reshape the drug discovery process.

Talking about genomics in drug development, **Dr Joydeep Goswami, Senior VP, Corporate Development, and Strategic Planning, Illumina** says, "Genomics will transform lifetime health management, improve outcomes and lower costs.

Probability of the drug molecule increases by 150 per cent when the drug target is supported by a genetically validated mechanism and cost per approval decreases by almost half, primarily through increased success rates. Genomics has the power to reshape drug discovery development.”

Cost-effective genome sequencing in India

India has seen an increase in genome sequencing in recent years. Though lagging behind China and Korea in the Asia Pacific region, a lot of Indian startups and companies have started undertaking research in this new technique. Government bodies are also increasingly recognising the importance of these advancements and turning to genomic research to provide personalised healthcare for patients.

Earlier in 2022, Singapore and India entered into an agreement to collaborate on genome and bioinformatics research, while the Genome Institute of Singapore has embarked on mapping Asian genomes to investigate how illnesses can affect different groups, providing valuable insight for researchers. The success of genomics innovation relies on robust and agile digital infrastructure to support the entire research programme, from data processing and exchange to the ability to analyse and store increasing data volumes.

Sharing his views on need for genome sequencing, **Murali Panchapagesa Muthuswamy, Chairman, Jananom; Chairman, Golden Jubilee Women's, Biotechnology Park and President, Council of Presidents of Association of Biotechnology Led Enterprise (ABLE)**, says, “We have to sequence a large number of genomes across ethnic groups among Indian populations and states from Northeast to South of India to get a decent representation. Credible bio markers and predictive and preventive models that are highly dependable will have to be developed. More efforts in this direction are needed and should be funded by government agencies since private bodies may not be able to afford 100k genome sequencing and million genome sequencing.”

With support and initiatives from the government agencies many startups have come up in the last few years and have been offering many services using genomic sequencing.

Bengaluru-based MedGenome has a state-of-the-art genetic lab where over 2,50,000 Exomes and Genomes are sequenced. It currently offers 1300 genetic tests and more than 9000 clinicians leverage the services. The company recently announced that it is offering Adaptive Biotechnologies' Next-Generation Sequencing (NGS)-based clonoSEQ Assay to assess minimal residual disease (MRD) in patients with multiple myeloma (MM), chronic lymphocytic leukaemia (CLL), and B-cell acute lymphoblastic leukaemia (B-ALL).

It has further unveiled VarMiner, an AI-enabled powerful variant interpretation software suite. The software will help clinicians, molecular geneticists, and genome analysts to interpret and report actionable variants. VarMiner supports various NGS Dx workflows like Germline Analysis where it covers all rare diseases, inherited cancers, Mitochondrial genome analysis, PGx and HLA analysis; Carrier/TRIO Analysis where it combines analysis of familial samples to detect De-novo and common inherited variants and reporting and somatic analysis, the comprehensive analysis of cancer genomes with support for liquid biopsy, haematology and solid tumour cases.

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Hyderabad-based Mapmygenome India has opened its Genomics Experience Centre in Bengaluru. The new centre will enable customers to get access to distinctive services such as genetic tests, genetic counselling, blood tests, health screening tests, antibody tests and RT-PCR test for COVID-19.

The company has launched a cost-effective pharmacogenomics solution MedicaMap. MedicaMap gives a detailed report about the body's response based on genetic makeup. It covers over 165 drugs across 12 specialities like cardiology, oncology, psychiatry, and many more. MedicaMap generates user-friendly, thorough reports on the toxicity and efficacy of medicines. Mapmygenome has also created several educational channels including Genomics Gupshup, blogs and conducts several workshops and CMEs.

Oncology startup 4baseCare has announced a collaboration with AstraZeneca India, a global science-led biopharmaceutical company for advancing cancer care in India. Under this collaboration, the two organisations will support advanced-stage cancer patients with targeted therapy options using affordable genomic solutions.

In another instance, C2i Genomics, a cancer intelligence company based in the US, has announced a strategic partnership with Mumbai-based startup Karkinos Healthcare, to co-develop the MRD market in India. The partnership enables C2i Genomics to bring innovative cancer detection technology to drive R&D, future pharma partnerships and clinical use in India and supports the company's long-term goals to scale pharma R&D projects around the globe. C2i Genomics' SaaS solution utilises a cloud-based platform to perform cancer tumour burden monitoring on a global scale, utilising equipped labs and sequencing networks worldwide.

Bengaluru-based Clevergene Biocorp, a deep-tech genomics company has signed a Memorandum of Understanding with the Rajiv Gandhi Centre for Biotechnology (RGCB), to establish a state-of-the-art genomics centre at Thiruvananthapuram. The state-of-the-art genomics centre will house the latest DNA sequencing equipment like the Illumina NovaSeq and other allied infrastructure to support cutting-edge research in plant biotechnology, infections, cancer and other chronic diseases. The services of this high-throughput genomics centre will be open for use in research to other institutes and industries and will

provide services like gene expression analysis, epigenetics and genome analysis including that of the COVID-19 virus.

Chennai-based LifeCell recently announced the addition of the 'NovaSeq 6000' sequencing system to their diagnostic technology platform. This development makes it one of the very few entities in India to have such exceptional capability which will enable more efficient and cost-effective genome sequencing. Following the successful implementation of Illumina's NovaSeq 6000 system, LifeCell will be able to leverage its technical expertise to improve scalability, support a varied range of applications, and simplify the workflow. Thus, this will provide high throughput speed and flexibility to carry out studies or testing which require processing a large amount of data, but in a faster and more economical way.

The company has also announced the launch of a revolutionary genetic test – Omega TB, in partnership with HaystackAnalytics, a genomics-based health-tech startup. The collaboration marks LifeCell's foray into the genetic testing industry for tuberculosis (TB) patients. The whole genome sequencing (WGS) test, designed by Haystack Analytics, aims to tackle the looming challenge of drug resistance. Apart from this, the test also helps in treating and preventing the spread of TB by providing a timely, affordable and accurate diagnosis.

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Bengaluru-based Strand Life Sciences, a genomics-based research and diagnostics company, has launched Strand Genomic Wellness, a new line of genomic-based tests for preventive wellness. Launched at Bengaluru Tech Summit this year, the new offering can help individuals understand and manage their disease better.

As the first offering of the programme, Strand has introduced the 'Genomic Health Insight' report to help individuals understand how their genomic variations might influence their risk for a broad range of diseases with 30-100 per cent heritability.

Researchers associated with the Tata Memorial Hospital in Mumbai and the Indian Institute of Technology, Bombay have developed India's first indigenously developed Chimeric Antigen Receptor (CAR) T-Cell therapy, a cutting-edge treatment for specific types of cancer patients. It has shown promising results. The therapy was tested on six paediatric patients of Acute Lymphocytic Leukaemia and 10 adults suffering from B-cell lymphoma as part of Phase I clinical trials by researchers. ImmunoACT along with Immuneel Therapeutics - backed by Biocon Biologics and top oncologist Dr Sidhartha Mukherjee - are two startups currently working to make CAR T-cell therapy available in India.

Recent launch of spatial transcriptomics and spatial proteomics have increased the confidence level of targeted therapies. Bengaluru-based TheraCUES Innovations has installed a technology platform from NanoString (GeoMx Digital Spatial Profiler (DSP) USA. Till a few years back, the cost of genome sequencing was huge. However, the costs have come down and are aiding researchers to take up more research activities.

Indigenisation, R&D and Governmental sops

Technological advancements in next-generation sequencing (NGS) and lower cheaper cost have led to an explosive growth of genomic data, both from clinical and research studies, according to **Ravi Gupta, Vice President – Bioinformatics, MedGenome Labs**. "The affordability of these new technologies has led to significant growth in mainstream diagnostics in human cancer and rare disorder diagnostics. The last decade has also resulted in several large population-level sequencing including 1000 genome, Genome Asia, gnomAD, TopMed, Icelander, UKBiobank, Japanese and many others. COVID-19 genome sequencing is one of the best examples of how the new genomics tools have changed our lives. The genomics data is astronomical and needs a huge computing and storage power for quick turnaround", he said.

Increasing economies of scale will reduce costs over time, but substantial R&D and indigenisation will be required to get costs to the level needed for very large-scale democratisation.

The pricing of gene therapies is a major issue in India. China and Korea have an advantage over India in volume processing. With the Indian government making the regulatory process much easier during the COVID-19 outbreak, a similar sop will be a win-win situation for the gene therapeutics market. Setting up labs and the availability of the right talent pools should be taken care of.

Anu Acharya, CEO, Mapmygenome, expounding on the cost-effectiveness of genome sequencing mentions, "The costs have come down dramatically from a few billion dollars to less than 100 dollars. That is more than what we saw in Moore's Law. Genomics has now become affordable and since germ-line mutations don't change over a lifetime, the value is very high. Streamlining new technologies, big data, and machine learning in genomic-based tests can have a massive economical impact on the healthcare system. With access to genome sequencing, an average consumer can reduce the odds of disease and enhance longevity most cost-effectively."

Says **Gopalakrishna Ramaswamy, Founder and CEO, TheraCUES Innovations**, "In a majority of developed countries, many genomic tests are paid by insurance/state health providers. However, in India, the patient has to pay. Though some insurance companies are bringing specific products based on disease, we need to have general acceptance for payment from insurance for tests, as it indirectly helps insurers in providing targeted therapies."

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Learning curve hurdles

Awareness seems to be the primary challenge around genomic tests. Once consumers and clinicians become aware of the value of genomic testing, they become strong advocates for them. Most genomic tests are deemed difficult to understand by clinicians and efforts must be undertaken to make the process easier and make the entire process actionable. Another challenge is in the space of reimbursement.

Speaking about challenges **Dr Ramesh Hariharan, CEO and Co-Founder, Strand Life Sciences**, says, “Consumer awareness on genomics is, of course, very low in India. There is also a lack of clarity on what genomics can or cannot help with. Genomics is a very high-complexity test with many nuances. Most tests take two or three days in the lab going through a multi-step process. That results in a lot of data that has to be analysed and placed in the context of available literature. This is very different from a biochemistry test where a sample goes into a machine and the result comes out. There are many parameters and many sources of error. Inadequate attention to these parameters can yield *garbage*.”

In a similar vein, **Ram Ramanujam, Founder & CEO, Propinquity Genomics**, says, “The main challenges in this industry now is that none of the equipment and reagents is made in India, and any NGS reagents made in India will reduce the pricing. One challenge that needs immediate attention is when you procure a new machine, and how to achieve RoIs in a quick time before the next model is released in the market. With evolving genomics enterprises in the world and emerging multi-scale biology and multi-omics with integrated biology, R&D must be incentivised in industry. As far as the R&D costs are concerned, sops from the Department of Biotechnology and Department of Scientific and Industrial Research (DSIR) will help new product development.”

The future

The future for genomics looks bright. In recent years, there has been steady progress in the advancement of newer sequencing technologies with never-before-seen features. Relying on able manpower, the right investments in research and timely government support, India is poised to set new benchmarks in the genomics space.

Talking about the growth of genomics in the country **Dr Shibichakravarthy Kannan, CEO, Oncophenomics** rightly points out, “You don't need a million dollars to start a genomics lab anymore. Anyone can start sequencing for as little as \$10,000. The democratisation of such newer sequencing technologies will facilitate more research programmes and fast-track translation into commercially viable products. We need to bridge the gap between academia and industry to make this happen. India has a unique advantage of skilled brain power. Bioinformatics training is now available free online and a readily available talent pool will further fuel the growth of the genomics industry in India.”

Speaking at the Bengaluru Tech Summit 2022 on opportunities for genomics research, **Dr Roger Hajjar, Head of Research and Development, Ring Therapeutics**, mentioned that the future looks bright for genomics in India as the country has a large patient population. India needs to establish a supply chain for raw materials. A large patient population in established tertiary centres is likely to kick-start any type of clinical therapy. The task ahead is enormous and there is a need to take stock of how much storage and processing speed is required. In this regard, more technological innovations in this space are expected to become game changers.

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